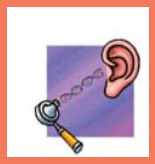
GENE NEWS

A Publication of the Hawai`i Department of Health Genetics Program

Early-Onset Hearing Loss Research Project... Approved!!!

Etiology of Hearing Loss Study



The Hawaii Genetics Program will be conducting a research project entitled "The Etiology of Congenital Hearing Loss," as reported in previous issues of Gene News.

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The Hawaii Genetics Program is pleased to announce that the study has been **approved** by the Institutional Review Boards (IRBs) of the Hawaii Department of Health, the University of Hawaii, and the Centers for Disease Control & Prevention (CDC)!

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IRBs are committees that review research involving humans to ensure that the research projects are ethical and of low risk to participants. The genetics evaluation and testing included in the Etiology of Congenital Hearing Loss study are often done in clinical genetics practice. However, this genetics follow-up and testing has not been offered routinely to those identified through newborn hearing screening. As such, this project is considered research and had to be approved by the appropriate IRBs before enrollment could begin.

How can referrals be made to the study?

Referrals to the study can be made by contacting one of the Genetic Counselors/ Study Coordinators for the project (contact information is listed on the right). If you have a child or patient who you think fits the criteria listed to the right, please contact us for more information!

Who can participate in the study?

The study is open to

- Children born in Hawaii on or after January 1st, 2001 who have been diagnosed with permanent hearing loss following a positive newborn hearing screen
- Children born in Hawaii on or after January 1st, 1998 who initially passed their newborn hearing screen, but were subsequently diagnosed with permanent hearing loss prior to their third birthday

The Genetics Program will be mailing study information to pediatric care providers across the State of Hawaii in the coming weeks. Information will also be posted on our website at:

www.hawaiigenetics.org

Please feel free to contact the Genetic Counselors for the Project:

Allison Taylor, MS 733-4998
allison@hawaiigenetics.org

Lianne Hasegawa, MS 733-9039
lianne@hawaiigenetics.org

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September, 2003

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Hawai'i Genetics Program

Hawai`i Department of Health
Children with Special Health Needs Branch

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Hawaii Community Genetics… Providing Care to Hawaii's Pediatric Genetics Population

IMPORTANT CONTACT INFORMATION

To Make a Patient Referral:

(808) 973-3403

Janet Brumblay,RN, MSN Genetics Nurse	973-3403
Susan Donlon, MS, CGC Genetic Counselor (QMC)	537-7633
Lianne Hasegawa, MS Genetic Counselor	733-9039
Christine Matsumoto,RN, MPH. Newborn Metabolic Screening Coordinator	733-9069
Allison Taylor, MS Genetic Counselor	733-4998
Greg Uramoto, MD Pediatric Endocrinologist	973-3403

In our June issue of Gene News, we announced the creation of **Hawaii Community Genetics**. Hawaii Community Genetics is a collaborative program involving the *Department of Health, Hawaii Medical Services Association, Kapiolani Medical Center for Women and Children, Queen's Medical Center,* and *the University of Hawaii John A. Burns School of Medicine*. Geneticists from the *School of Medicine* visit Hawaii approximately one week per month for pediatric genetics clinics, as well as to provide genetics education to various groups.

The first Hawaii Community Genetics clinic took place the week of June 23rd, 2003. Dr. Eugene Hoyme (Geneticist), Dr. Greg Enns (Geneticist) and Susan Schelley (Genetic Counselor) visited from Stanford to join Hawaii-based health care providers (contact information is to the left). The Stanford team provided direct clinical services for the June patients, and were supported by the efforts of many others, including Gayle Kearney, RN, and Karen Uehara, Manager of Hawaii Community Genetics. During the week of June 23rd, 27 patients were evaluated in clinic, and several more patients were seen as in-hospital consults at Kapiolani Medical Center for Women and Children.

During the July Hawaii Community Genetics clinic, 21 patients were seen, as well as several more in-hospital consults. Dr. Greg Enns attended from Stanford, and joined the same Hawaii-based team to provide services in July.

The June and July clinics ran very smoothly, with many families reporting much appreciation for and satisfaction with the services. Unfortunately, due to circumstances beyond our control, the August genetics clinic had to be cancelled. We apologize for any inconvenience this may have caused the families or referring physicians. We are working to increase the length of a future clinic to make up for the cancelled August clinic. We are also investigating the use of telemedicine to provide more consultation opportunities for families. We appreciate the community support for our collaborative effort to provide clinical genetic services.

Coordinator's Corner

You may have noticed that GeneNews has expanded from four to eight pages! We have so much information to report that we've had to move to the larger format. You can tell that the Genetics Program has been very busy! Besides the expansion of GeneNews, we are also excited about our new website renovation based on user feedback (more information on the next page). We really appreciate all the users that have given us comments about improving the website to make it more useful for you.

I really like to hear from our readers. If you have comments (good or bad) or questions about GeneNews, our website or any of our activities, please contact me at **sylvia@hawaiigenetics.org** or **733-9063**. I look forward to hearing from you.

Sylvia M. Au, M.S., C.G.C.
State Genetics Coordinator

Survey results are in!

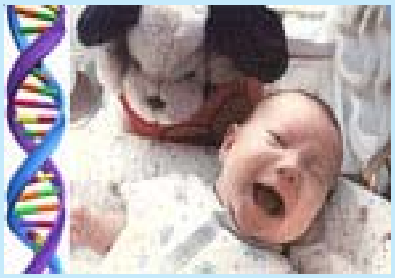
On April 12, 2003, the Hawaii Science Teachers' Association (HaSTA) held a half day conference at Punahou School. The conference was open to public and private science teachers statewide. The DOH Genetics Program surveyed the teachers regarding genetics in their curriculum in preparation to create a "Genetics Resource Kit" which is intended to aid both junior high and high school science instructors in teaching genetics.

On average, high school and junior HS science and biology teachers reported spending approximately 20 hours per year teaching genetics. The survey found that many teachers cited a lack of genetics resources for teaching, as well as difficulty in keeping up-to-date with current developments in genetics, to be among the most common barriers to teaching genetics. In light of these barriers, many teachers indicated that they would like the opportunity to increase genetics education in their classrooms. 72.8% expressed interest in having a genetics guest speaker come to their class, and 76.2% are interested in having access to a traveling genetics lab. Interest in the Genetics Resource Kit was quite positive as well, with 72.6% of respondents indicating that they would like to contribute to or provide feedback for their development.

We will be contacting teachers shortly for assistance in the development, testing and implementation of these teaching resource kits.

The DOH is in the initial stages of preparing Genetics Resource Kits for genetics education. If you did not participate in our survey, but would like to contribute to their development, please email us at:

teachingtools@hawaiigenetics.org



www.hawaiigenetics.org

Genetics Website Revamped!

Comments from Hawaii Genetics Program website users have indicated that because there is so much information on the site, some of it may be difficult to find! In light of these comments, the Hawaii Genetics Program conducted a revision of the website, led by **Amanda Tice**, summer intern from the University of Chicago. **Nicole Sameit** of the Hawaii Genetics Program had done a number of interviews previously with individuals regarding the website. To follow up on the interviews, Amanda devised a website user survey to determine what type of information people are most interested in while browsing the genetics site. The survey results were used to help build a new site map, which will organize information on the website into a more user-friendly order. Additionally, a new "quick start" menu will be added to allow users to jump directly to the specific information they want. The new Hawaii Genetics Program website is expected to be up and running by the end of September.

Be sure to check it out!

Launch of Expanded Newborn Metabolic Screening Statewide

~~ Using Tandem Mass Spectrometry~~

LIST OF EXPANDED NEWBORN SCREENING DISORDERS

1. Congenital Hypothyroidism
2. Congenital Adrenal Hyperplasia (CAH)
3. Biotinidase Deficiency
4. Hemoglobinopathies

Amino Acid Disorders
5. Homocystinuria
6. Phenylketonuria (PKU)
7. Tyrosinemia (Types I and II)
8. Galactosemia
9. Maple Syrup Urine Disease (MSUD)

Urea Cycle Disorders
10. Arginase deficiency
11. Argininosuccinate lyase deficiency (ASA)
12. Citrullinemia

Organic Acidemia Disorders
13. Beta-ketothiolase deficiency
14. Glutaric acidemia, Type I
15. Isobutyryl CoA dehydrogenase deficiency
16. Isovaleric acidemia
17. Malonic aciduria
18. Methylmalonic acidemias (MMA-8 types)
19. Multiple carboxylase deficiency
20. Propionic acidemia
21. 2-Methyl-3-hydroxybutyryl CoA dehydrogenase deficiency
22. 2-Methylbutyryl CoA dehydrogenase deficiency
23. 3-Hydroxy-3-methylglutaryl (HMG) CoA lyase deficiency
24. 3-Methylcrotonyl CoA carboxylase deficiency (3-MCC)
25. 3-Methylglutaconyl CoA hydratase deficiency

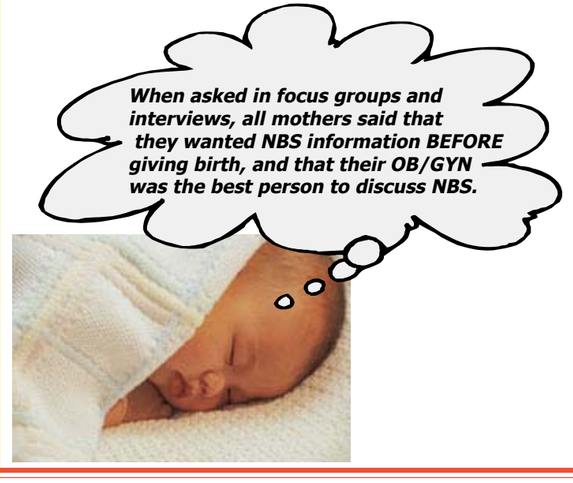
Fatty Acid Oxidation Disorders
26. Short chain acyl-CoA dehydrogenase deficiency (SCAD)
27. Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
28. Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
29. Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
30. Multiple acyl-CoA dehydrogenase deficiency (MADD)
31. Carnitine uptake/transport defects

What Are You Talking About?

Newborn metabolic screening(NBMS) is performed when a baby is just a few days old. While still in the hospital, the baby's heel is pricked, and a few drops of blood are collected. This blood is then sent to a laboratory and tested to see if the baby has a metabolic disorder. Currently, all babies born in Hawaii are tested for seven disorders. However, thanks to a new technology called tandem mass spectrometry, all newborns can now be tested for thirty-one disorders with the same amount of blood. Newborn screening is important because the earlier a baby with a metabolic disorder is diagnosed, the earlier treatment can begin, and the better the chance of a normal, healthy life.

How Will This Affect Me if I am a Health Care Provider?

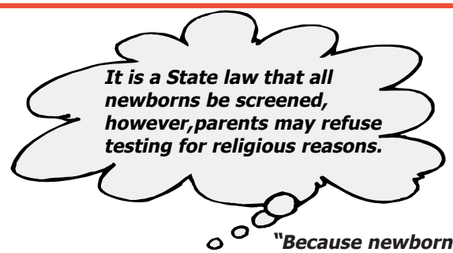
The change in the number of disorders in the newborn screening panel will not affect your role and your responsibilities. Newborn blood is collected in the same way, and you will still be responsible for informing parents of their child's newborn screening results. Should one of your patients have a initial positive result, the Newborn Screening Program, Hawaii medical consultants, and laboratory consultants will all work with you to provide your patient with the proper follow-up testing, counseling, and support.



Extra! Extra!

Read all about it!

On **September 1, 2003**, Hawaii expanded the number of disorders included in its mandatory newborn metabolic screening panel…from seven to **thirty-one!**



"Because newborn metabolic screening is a screening test, a initial positive result does not mean necessarily that the baby has a metabolic disorder. More testing is needed to confirm the diagnosis, and parents should bring their baby back for testing as soon as possible."

How Do You Know That Expanded Newborn Screening Works?

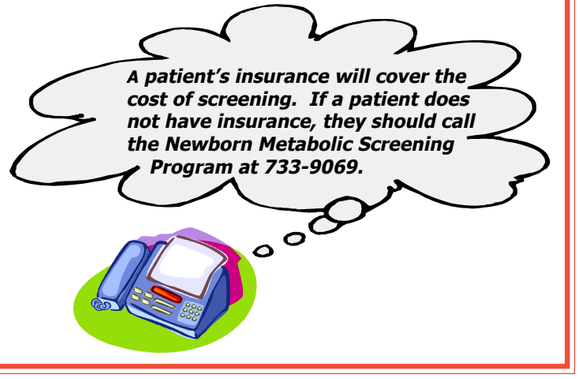
Since March 1, 2002, Hawaii and California have collaborated on an expanded newborn screening pilot project. As part of this project, the parents of all babies born at Kapiolani Medical Center were offered expanded newborn screening. During the project, over 300,000 babies were screened in Hawaii and California. Of those screened, over 500 received initial positive expanded newborn screening results, and 48 of these babies were confirmed to have metabolic disorders. In Hawaii, six babies received initial positive expanded newborn screening results, and one baby were confirmed to have a metabolic disorder called isovaleric acidemia. With the knowledge of this diagnosis and the immediate start of treatment, the severe mental retardation which otherwise would result will most likely be prevented. Thanks to the pilot project, the baby is now eleven months old and doing well!

How Will This Affect Me if I am a Parent?

No additional blood is needed from your baby's heel than that which is already being taken for the seven disorders. However, your baby will benefit from being screened for thirty-one disorders! Your doctor should tell you the results of your baby's screening test. If your baby does have an initial positive or questionable test result, it does not necessarily mean that your baby has a metabolic disorder. It means that more testing must be done to confirm whether or not your baby actually has a disorder. So, if you are asked to have some follow-up tests on your baby, please have them done as soon as possible!

How Can I Get More Info?

For more information about newborn screening in Hawaii, call the State Newborn Metabolic Screening Program at **(808)733-9069** or go to **www.hawaiigenetics.org** or **www.newbornscreening.info**. The www.newbornscreening.info website, which is dedicated to information about expanded newborn screening, has a section for families as well as one for health care providers. The website also includes a general genetics overview, a description of tandem mass spectrometry, a glossary, and detailed fact sheets on each of the metabolic disorders screened for in expanded newborn screening. The fact sheets, which were written by a genetic counselor and reviewed by metabolic geneticists, have information such as the incidence, symptoms, natural history, and treatment of the disorders.



2nd Annual Global Public Health Conference

The second annual Global Public Health Conference was held June 4th-6th, 2003 at the Hawaii Convention Center. This conference attracted a wide range of public health attendees, representing a number of professions and locations across Hawaii, the Pacific Islands, and beyond.

The Hawaii Genetics Program had an information booth and Program staff present to answer questions about the program and its activities. Conference attendees who completed a short survey were given our new custom-made Genetics Program word magnets. In addition, the State Genetics Coordinator gave a workshop presentation on Expanded Newborn Metabolic Screening and another workshop presentation on Integrating Genetics into Public Health.

The short survey was completed by a total of 90 attendees, and was intended to assess demographic information, knowledge of and opinions regarding genetics, and perspectives of genetics in public health. See Figure 1 for demographic information.

Figure 1: Where did the respondents come from?

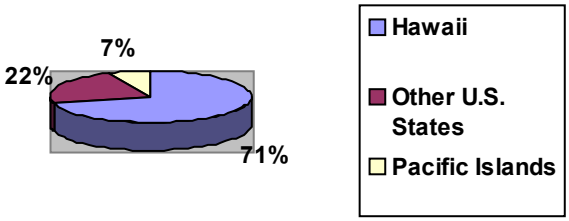
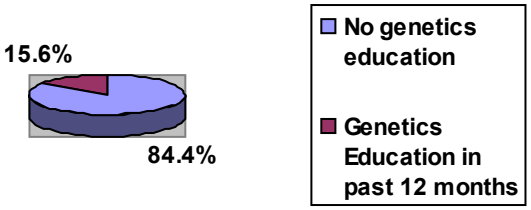


Figure 2: Have You Obtained Genetics Education in the Past 12 months?



When asked about previous genetics education, many (56.1%) reported receiving their most recent formal genetics education in college or graduate school. 16.7% reported their most recent formal genetics education as being obtained through conferences. ***In terms of obtaining any genetics education, only 15.6% reported not receiving any genetics education in the past 12 months (Figure 2).***

Caffeine Free

This may be just the answer for those of us who love the taste of coffee, but want to cut out the caffeine!

The traditional decaffeination process uses water or organic solvents to remove caffeine as the beans roast. Unfortunately, this process also removes some of the flavor and aroma of the final product. Through genetic modification, scientists have been able to interfere with the gene responsible for triggering caffeine production. Preventing the actual caffeine production in the plant would eliminate the need for chemical decaffeination. So, we would expect this caffeine-free coffee to maintain the taste of regular coffee.

Figure 3 shows the methods used to obtain genetics education. These statistics support the Hawaii Genetic Program's efforts to increase genetics education both at appropriate conferences and through our website (www.hawaiigenetics.org).

Figure 3: How is Genetics Education Obtained?

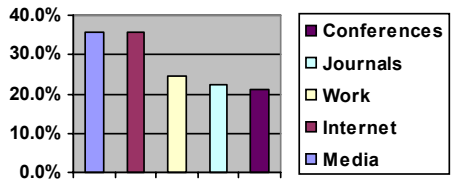
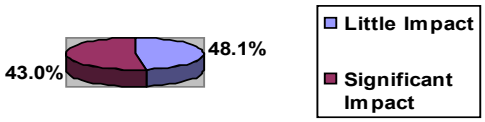


Figure 4: Current Impact of Genetics



Survey respondents were asked their perspectives of the impact genetics currently has and will have on public health programs. Genetics having a current impact on public health programs was perceived by 91.1% of respondents overall (Figure 4).

When asked about their opinion of the impact genetics will have on public health programs in the next ten years, 98.6% supported that genetics will have an impact (Figure 5).

Figure 5: Impact of Genetics in the Next 10 Years

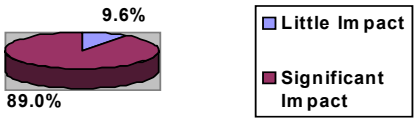
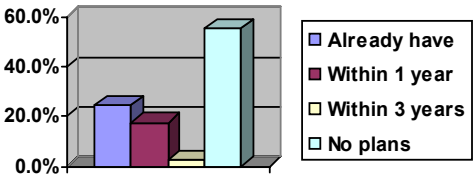


Figure 6: Plans to Incorporate Genetics into Their PH Programs?



The survey also questioned whether genetics services or education is or will be included in the respondents' program activities. Figure 6 shows their plans to incorporate genetic services or education into their public health programs. Perhaps disappointingly, 55.3% reported no plans to incorporate genetics into their program activities. One of the Hawaii Genetics Program goals is to increase the number of public health programs that incorporate genetics into their program and we will continue to recruit and collaborate with new programs towards this goal.

Since Hawaii is expanding its newborn screening program, we wanted to assess respondents knowledge of newborn metabolic screening. **All** respondents were correct in answering that the states and territories in the US do not all screen for the same disorders. However, the other five newborn screening questions were answered correctly by only 51.1% or less of the respondents.

The final knowledge question on the survey was intended to assess respondents' awareness of whether predictive genetic testing is available for certain conditions. The majority of respondents (61.1%-88.9%) were correct in responding that predictive testing is not available for alcoholism, obesity, intelligence, and autism. On the flip side, for two conditions for which predictive testing is available (hereditary breast cancer and colon cancer), only 42.2% and 38.9% (respectively) were aware of this.

Interested in reading more? Check out our website (www.hawaiigenetics.org) for a more detailed report of the surveys from the Global Public Health 2003 Conference.

Coffee Beans?

Researchers have been attempting to get a genetic jump on the decaffeination process here in Hawaii. Waialua-based *Integrated Coffee Technologies, Inc.* may have these "less caffeinated" coffee plants ready for trial in the next 2-3 years.

Although sales of decaffeinated coffee are expected to rise in the coming years, it is difficult to predict exactly how the public will embrace this novel product since genetically-modified foods remain an issue of controversy.

For more information, please go ***www.hawaiigenetics.org***

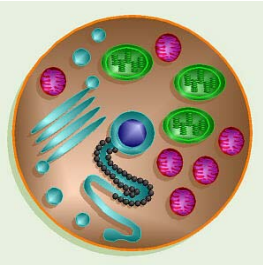
HYPOMAGNESEMIA

The identification of a genetic defect that leads to the disease known as hypomagnesemia was reported recently by two international groups of researchers (*Nature Genetics 31: 166-174*). Hypomagnesemia, also known as HSH (*OMIM 602014*), is characterized by extremely low levels of magnesium and calcium in serum. Both magnesium and calcium play critical roles in a wide range of cellular functions, and the low levels found in people with this disorder (especially infants) can result in a number of neurological problems such as seizures and muscle spasms.

Hypomagnesemia is rare in the general population, but it can occur at higher frequencies among people of Arabic descent. Three Bedouin kindreds studied, for example, contained 11 males and two females that were affected. The preponderance of affected boys in these and other families had previously led researchers to believe that the gene responsible was X-linked. The recent studies, however, have clearly shown that the defect is due to an autosomal gene on chromosome 9 (location: 9q22), and that the mutation is recessive.

The mutated gene the researchers identified normally produces a protein involved in control of membrane potential and the movement of ions in and out of cells. This finding is completely consistent with previous evidence showing that the primary physiological problem associated with HSH was a defect in intestinal magnesium transport.

The identification of this gene also has implications beyond an improved understanding of the genetic basis of this disorder. Recent studies have shown, for instance, that up to 60% of patients in intensive care units may suffer from an induced form of hypomagnesemia, possibly due to the administration of intravenous fluids. A number of drugs with diuretic effects may also lead to similar problems. The researchers are hopeful that the identification of this gene may lead to better insights into the physiology of magnesium transport for these and other potential applications.



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The Health State
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